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Newborn screening in metabolic Diseases

In the early 1960s, Robert Guthrie and Ada Susi published a method for the detection of phenylketonuria (PKU) in newborns. A small punch from a blood or urine spot on a filter paper card was applied to an agar plate. Elevated phenylalanine (Phe) levels impaired the growth of Bacillus subtilis ATCC 6051, and the diameter of clearing of bacterial growth could be correlated with Phe levels in the blood spot. Screening programs were rapidly developed in the industrialized world, and the early detection and treatment of PKU has led to dramatically improved, and sometimes normal, outcomes. This method for screening, known as a bacterial inhibition assay, was employed to detect other inborn errors of metabolism such as galactosemia, maple syrup urine disease (MSUD), homocystinuria, and others, though they were not as widely adopted as screening for PKU. Enzyme assays were subsequently developed for newborn screening (NBS) bloodspots for detection of disorders, including galactosemia in 1962 and biotinidase in 1984. In 1975, an immunoassay procedure was published for the detection of neonatal hypothyroidism and over the next few decades immunoassays were developed for congenital adrenal hyperplasia (21-hydroxylase deficiency) and other disorders. In the 1980s and 1990s, fluorometric assays replaced bacterial inhibition assays for analyte analysis. In 1990, tandem mass spectroscopy (MS/MS), which had been used clinically to measure urine acylcarnitine's, was demonstrated to be amenable to the detection of analytes in NBS bloodspots. The adoption of this methodology by NBS programs in North America and in most industrialized nations rapidly expanded the number of disorders included in NBS programs. This rapid expansion reflected the large number of analytes that could be detected with a single assay; the high level of automation; and the speed of the sample preparation, assay, and analysis. In 2006, the American College gave future directions to the rapid improvement and reducing costs of many high throughput technologies, the expansion of disorders included in NBS programs worldwide is highly likely. Over the past few years, many conditions have been under active investigation for the feasibility and validation of high throughput population screening. The list includes disorders of creatine metabolism, glucose-6-phasphate dehydrogenase deficiency, various lysosomal storage disorders, severe combined immunodeficiency, disorders of sterol metabolism (particularly Smith-Lemli-Optiz syndrome), Wilson disease, X-linked adrenoleukodystrophy, familial hypercholesterolemia, Fragile X syndrome, Duchene muscular dystrophy, and spinal muscular atrophy. In addition to feasibility, the principles described in Section II of this chapter should be applied in the decision regarding whether to add a particular disorder to NBS programs. Finally, especially as the cost of next-generation sequencing technologies continues to plummet and as our ability to accumulate and analyze large DNA sequence data sets continues to improve, it is intriguing to consider the future possibility of whole genome or whole exome sequencing as an additional or even replacement technology for NBS.

Biography

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